Your Guide to Understanding Genetic Conditions

FZD6 gene

frizzled class receptor 6

Normal Function

The *FZD6* gene provides instructions for making a protein called frizzled-6. This protein is embedded in the outer membrane of cells, where it is involved in transmitting chemical signals from outside the cell to the cell's nucleus.

Specifically, frizzled-6 participates in the Wnt signaling pathway, a series of steps that affect the way cells and tissues develop. Wnt signaling is important for cell division (proliferation), attachment of cells to one another (adhesion), cell movement (migration), and many other cellular activities. At the cell surface, frizzled-6 attaches (binds) to certain proteins, which triggers the frizzled-6 protein to send signals into the cell and initiate the Wnt signaling pathway.

Frizzled-6 is active in many tissues. During early development it plays a critical role in the growth and development of nails, particularly the attachment of the nail to the nail bed. Studies suggest that frizzled-6 is also involved in hair growth.

Health Conditions Related to Genetic Changes

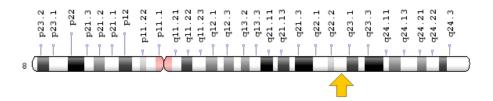
nonsyndromic congenital nail disorder 10

At least three mutations in the *FZD6* gene have been found to cause nonsyndromic congenital nail disorder 10. This condition is characterized by abnormally thick fingernails and toenails that may appear claw-like. The mutations lead to the production of a frizzled-6 protein that cannot get to the cell membrane where it is needed or that cannot transmit signals into the cell. As a result, Wnt signaling is disrupted and the growth and development of nails is poorly regulated, which leads to separation of the nail from the underlying nail bed (onycholysis) and other abnormalities of the fingernails and toenails.

Chromosomal Location

Cytogenetic Location: 8q22.3, which is the long (q) arm of chromosome 8 at position 22.3

Molecular Location: base pairs 103,298,433 to 103,332,866 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- frizzled 6, seven transmembrane spanning receptor
- frizzled family receptor 6
- frizzled homolog 6
- FZ-6
- FZ6
- HFZ6

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): The Wnt Pathway https://www.ncbi.nlm.nih.gov/books/NBK10043/#A1061
- Madame Curie Bioscience: Secreted Antagonists/Modulators of Wnt Signaling https://www.ncbi.nlm.nih.gov/books/NBK6536/
- Molecular Biology of the Cell (fourth edition, 2002): Wnt Proteins Bind to Frizzled Receptors and Inhibit the Degradation of β-Catenin https://www.ncbi.nlm.nih.gov/books/NBK26918/#A2887

Scientific Articles on PubMed

PubMed

https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FZD6%5BTIAB%5D%29+OR+%28frizzled-6%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

 FRIZZLED, DROSOPHILA, HOMOLOG OF, 6 http://omim.org/entry/603409

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology http://atlasgeneticsoncology.org/Genes/GC_FZD6.html
- ClinVar https://www.ncbi.nlm.nih.gov/clinvar?term=FZD6%5Bgene%5D
- HGNC Gene Family: G protein-coupled receptors, Class F frizzled http://www.genenames.org/cgi-bin/genefamilies/set/286
- HGNC Gene Symbol Report http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/ hgnc_data.php&hgnc_id=4044
- NCBI Gene https://www.ncbi.nlm.nih.gov/gene/8323
- UniProt http://www.uniprot.org/uniprot/O60353

Sources for This Summary

- OMIM: FRIZZLED, DROSOPHILA, HOMOLOG OF, 6 http://omim.org/entry/603409
- Fröjmark AS, Schuster J, Sobol M, Entesarian M, Kilander MB, Gabrikova D, Nawaz S, Baig SM, Schulte G, Klar J, Dahl N. Mutations in Frizzled 6 cause isolated autosomal-recessive nail dysplasia. Am J Hum Genet. 2011 Jun 10;88(6):852-60. doi: 10.1016/j.ajhg.2011.05.013. Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21665003

 Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3113248/
- Guo N, Hawkins C, Nathans J. Frizzled6 controls hair patterning in mice. Proc Natl Acad Sci U S A. 2004 Jun 22;101(25):9277-81.

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15169958
Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC438967/

- Naz G, Pasternack SM, Perrin C, Mattheisen M, Refke M, Khan S, Gul A, Simons M, Ahmad W, Betz RC. FZD6 encoding the Wnt receptor frizzled 6 is mutated in autosomal-recessive nail dysplasia. Br J Dermatol. 2012 May;166(5):1088-94. doi: 10.1111/j.1365-2133.2011.10800.x. Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22211385
- Raza SI, Muhammad N, Khan S, Ahmad W. A novel missense mutation in the gene FZD6 underlies autosomal recessive nail dysplasia. Br J Dermatol. 2013 Feb;168(2):422-5. doi: 10.1111/j.1365-2133.2012.11203.x.

Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22861124

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